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Family Amaurotic Idiocy without Characteristic Ophthalmoscopic Signs.

By F. PARKES WEBER, M.D.

THE child, M. A., female, aged 14 months, is not wasted, but is unable to sit up, and, owing to weakness in the muscles of the back and neck, its head (which seems rather large and heavy, and measures 18½ in. in circumference) falls backwards if not supported ("head-lolling"). There is occasionally likewise active retraction of the head, which must not be confused with the merely passive head drop. It cannot move about; it cannot even turn or roll itself over in the bed from one side to the other. It can, of course, only be fed with fluids, and when it sucks from a feeding-bottle the movements of its jaws appear automatic, rhythmic, and exaggerated; they suggest the automatic movements of a frog deprived of its cerebrum. Similar rhythmic movements of the jaws and mouth are sometimes present when the child is not feeding. It occasionally "swallows the wrong way." It generally lies apathetic or somnolent, taking no notice of anything unless disturbed by medical examination or otherwise, but will cry out when the feeding-bottle is removed. It never attempts to grasp or even to touch anything, not even the feeding-bottle. There is a variable amount of rigidity, due to tonic muscular spasm, affecting the trunk and both upper and lower extremities. The lower extremities are often rigidly extended, with the feet in a tetany-like position; the upper extremities are flexed at the elbows, and the hands are often clenched, with the thumbs between the middle and index fingers. The knee-jerks can be obtained, and are at present rather exaggerated. No ankle clonus. The plantar reflexes tend, on the whole, to be of the extensor type

(Babinski sign). The tendon reflexes of the upper extremities are present. There is vertical nystagmus, which varies much in degree from time to time: sometimes it is very well marked, rendering ophthalmoscopic examination difficult; sometimes it is temporarily absent.

Ophthalmoscopic examination (Dr. R. Gruber and Dr. C. Markus) shows nothing characteristic of family amaurotic idiocy. There is no pallor of the macular region with a cherry-red spot (corresponding to the fovea centralis) in the centre, such as was described first by Waren Tay in 1881, and by B. Sachs in 1887, and has since been found to be present in nearly all (some say all the certain) cases of the disease.[1] Both optic disks are rather pale, but not distinctly atrophic, and there is no evidence of past optic neuritis. The child is practically blind, though it can still perceive a bright light, and will occasionally follow an electric lamp with its eyes. The pupils react promptly to light.

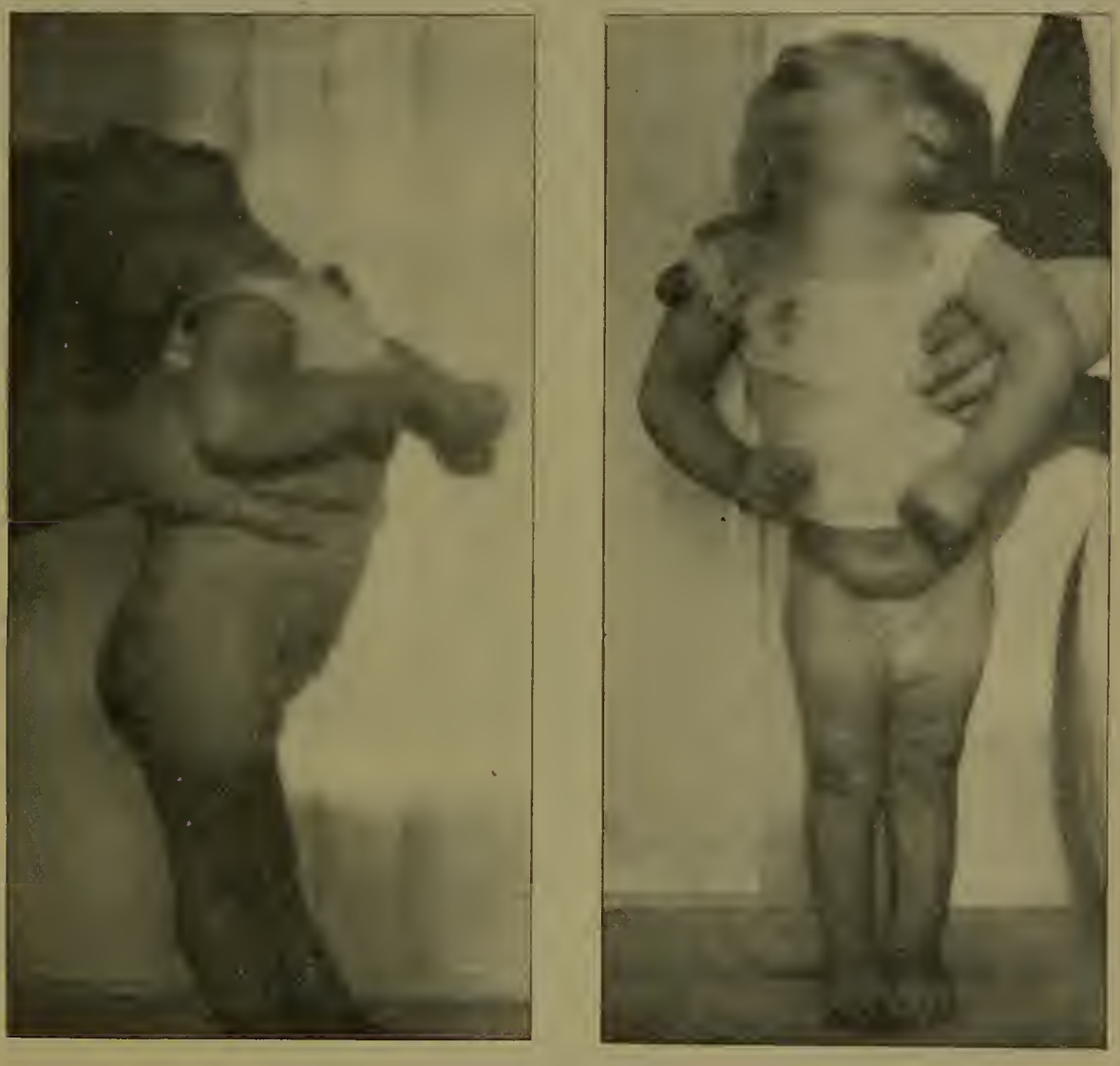
There seems to be nothing abnormal in regard to cutaneous sensation, hearing, or taste. The child notices at once if its milk is unsweetened. The only teeth visible are the lower central incisors; there are no signs of rickets at the ribs or epiphyses. There is no evidence of disease in the abdominal or thoracic viscera, excepting some bronchitis, possibly connected with "swallowing the wrong way." Lately there has been variable fever. The urine is free from albumin and sugar.

The disease has been a very chronic one. According to the mother, the child was normal at birth and was suckled for the first five months. Weakness in the muscles of the back and neck, so that it could not hold up its head, was observed at three months of age. The mother does not know whether it could ever see properly. There is no history of convulsions or any illnesses other than the present one.

The parents, both of them said to be healthy, are Jews, but there is no blood-relationship (consanguinity) between them. They have had six children, of which the present patient is the youngest. The mother says she has had no miscarriages. The eldest child is a girl, now aged 11 years, living and healthy. The second, a boy, is likewise living and healthy, aged 9½ years. The third, a girl, had the same disease, so at least the mother thinks, as the present patient, and died at one year of age in the

[1] E. C. Kingdon and Risien Russell, in their classical paper (*Med.-Chir. Trans.*, Lond., 1897, lxxx, p. 106), say: "The ocular symptoms, which are an early and, we believe, absolutely diagnostic sign of the disease, consist in symmetrical changes at the macula lutea, in which situation, covering a space nearly twice the size of the optic disk, is a whitish-grey patch. . . . In the centre of the patch is seen the fovea centralis as a dark cherry-red spot."

mother's home; there was no necropsy. The fourth child was a boy (Ben A.), who likewise, according to the mother, suffered from the same disease; at the age of 2 years he was admitted with convulsions to the London Hospital on June 2, 1906, and died on the following day. Dr. H. L. Tidy, the Medical Registrar of the London Hospital, has kindly furnished the following notes of the case: The head was $20\frac{3}{4}$ in.



Photographs showing the head-drop and the position of the extremities.

in circumference, and the forehead large and square. There were no marked epiphysial signs of rickets. The boy had been healthy at birth and had been breast-fed for the first twelve months. He had never been able to take solid food. The head had been inclined backwards since three months of age. He had had convulsions every few weeks since six months old. He had never been able to walk or talk. On admission

to the London Hospital the boy was quite stiff, the arms in a tetany-like position, and the head thrown back. There was a patch of pleural friction on the right side; afterwards the rigidity passed off and the child slept. Temperature, 103·6° F. At the necropsy the lungs showed areas of collapse, and the larynx contained vomit. A large pad of adenoids was found in the nasopharynx. There was internal hydrocephalus. The skull-cap was thin and the fontanelle was widely open. The brain weighed 54 oz., and the brain-substance was soft.

The fifth child, a male, appeared healthy at first, but died of bronchitis at the age of eleven months, in the mother's home. There was no necropsy.

Dr. Weber thinks that the present case is almost certainly one of family amaurotic idiocy, possibly associated with internal hydrocephalus. The interesting point in the present case is that, as the typical ophthalmoscopic appearances of the disease are absent, and as there is no atrophy of the optic disks, it is unlikely that the amaurosis is connected either with local changes in the retina or with changes secondary to hydrocephalus (even if there be any hydrocephalus). The amaurosis is, therefore (in the present case), probably dependent on the diffuse changes (atrophy of ganglion cells and fibres) such as have been demonstrated in the brains of cases of family amaurotic idiocy in which during life the characteristic ophthalmoscopic changes were present. There is no a priori reason why the ganglion cells of the retina, the atrophy of which is supposed to give rise to the ophthalmoscopic changes in question, should be affected in every case of the disease. Indeed there are several records [1] suggesting that the retinal changes, although they led to the discovery of the disease, are not an altogether essential part of the pathological process. Unfortunately necropsy records of such clinically "incomplete" cases are wanting, so that it has not yet been proved that the central nervous system in the "incomplete" cases shows changes exactly similar to those which have been proved to be present in "clinically" complete cases. Many of the clinical signs (spasticity of limbs, peculiarity in sucking, and difficulty in swallowing) in the present case, as in typical cases, are doubtless really due to a kind of chronic cerebral diplegia produced by the morbid process; and other signs can be explained by the cerebellum being affected also in the same way as the cerebrum.

[1] See the cases quoted from the literature of the subject by Wilbrand and Saenger, *Die Neurologie des Auges*, Wiesbaden, iv, part 1 (1909), pp. 396—447. Moreover, in the so-called "juvenile type" of the disease the characteristic ophthalmoscopic changes are not found.

Internal Hydrocephalus and Amaurosis without Definite Ophthalmoscopic Changes, following Symptoms of Posterior Basic Meningitis or Ependymitis.

By F. Parkes Weber, M.D.

The patient, H. H., a boy, was aged 4½ months when admitted to the German Hospital on October 11, 1909. He had seemed healthy until eight days before admission, when he commenced to suffer from slight convulsions, spasmodic movements of eyes, and diarrhœa. There was nothing of special interest in the family history, except that a brother of the patient, aged 11 years, suffered from congenital hydrocephalus. Since admission to the hospital the child has seemed almost blind, and has become increasingly apathetic, although at first there was considerable irritability. There have been no convulsions, excepting a slight one when the child was being bathed directly after admission. Decided retraction of the head was at first very noticeable, but is less marked at present. The head has gradually increased in size. Its circumference on November 8, 1909, was 18⅛ in., and on January 24, 1910, was 20⅝ in. At first there was slight fever, but, excepting during three days in January, there has been none since October 21. At present the child is practically blind, though it can perceive a bright light. Ophthalmoscopic examination shows nothing abnormal beyond slight pallor of the optic disks. The child lies quite apathetic in its bed, not moving from the position in which it is placed. It never attempts to catch hold of anything, and does not even put its hands towards the feeding-bottle. It can distinguish between sweetened and unsweetened milk, and can evidently hear a loud, sudden sound. There is often considerable spasticity of the lower extremities, on which occasions (though not always on other occasions) the plantar reflexes are of the extensor type (Babinski's sign).

Splenomegaly and Hydrocephalus.

By F. Parkes Weber, M.D.

The patient, W. J., a boy, was aged 13 months when admitted to the German Hospital in August, 1909. He was very pale, with a large head, measuring $20\frac{7}{8}$ in. in circumference, and an open anterior fontanelle of about the size of a shilling. There was doubtless some internal hydrocephalus. The spleen was enlarged, and reached nearly to the ilium. The liver extended three finger-breadths below the costal margin. The urine was free from albumin and sugar. Examination of the blood showed: Hæmoglobin, 29 per cent. (Sahli's method); red cells, 2,568,000 in the cubic millimetre of blood; white cells, 6,300 (neutrophile polymorphonuclears 32 per cent., small lymphocytes 33 per cent., large lymphocytes 24 per cent., eosinophiles 1 per cent., basophiles 6 per cent.); no nucleated red cells were seen. Although there was no history of congenital syphilis, the treatment adopted has been chiefly anti-syphilitic. The child's condition has slowly but decidedly improved. Examination of the blood on January 22, 1910, showed: Hæmoglobin, 40 per cent. (Sahli's method); red cells, 4,480,000 in the cubic millimetre of blood; white cells, 13,200 (neutrophile polymorphonuclears 43·2 per cent., small lymphocytes 44·6 per cent., large lymphocytes 4·8 per cent., transitionals 3·4 per cent., eosinophiles 1·2 per cent., basophiles 2·8 per cent.); during the count of 500 white cells, 11 nucleated red cells were observed. The circumference of the head is now $21\frac{1}{4}$ in., and the fontanelle has almost closed. The spleen hardly reaches below the umbilical level, and the liver cannot be felt below the costal margin in the right nipple line. Beyond moderate beading of the ribs, the child has not shown marked signs of rickets. There was slight pyrexia on admission, but otherwise the child has generally been free from fever. Wassermann's sero-reaction for syphilis, kindly tried by Dr. Henderson Smith on November 28, 1909, gave a negative result.

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